MMAB gene

methylmalonic aciduria (cobalamin deficiency) cblB type

Normal Function

The *MMAB* gene provides instructions for making an enzyme that is involved in the formation of a compound called adenosylcobalamin (AdoCbl). AdoCbl, which is derived from vitamin B12 (also known as cobalamin), is necessary for the normal function of another enzyme known as methylmalonyl CoA mutase. This enzyme helps break down certain proteins, fats (lipids), and cholesterol.

The MMAB enzyme is active in mitochondria, which are specialized structures inside cells that serve as energy-producing centers. Once vitamin B12 has been transported into mitochondria, the MMAB enzyme converts a form of the vitamin called cob(I)alamin to AdoCbl. Studies suggest that this enzyme may also deliver AdoCbl to methylmalonyl CoA mutase.

Health Conditions Related to Genetic Changes

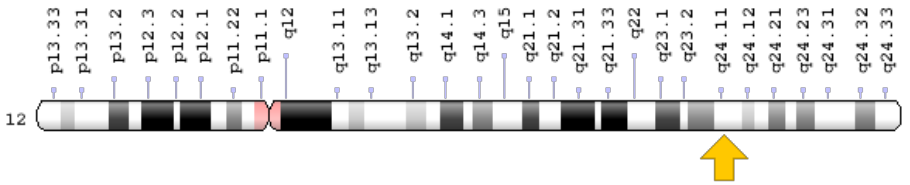
methylmalonic acidemia

At least 25 mutations in the *MMAB* gene have been found to cause methylmalonic acidemia, a condition characterized by feeding difficulties, developmental delay, and long-term health problems. Some of these genetic changes delete or duplicate a small amount of genetic material in the *MMAB* gene. Other mutations change a single protein building block (amino acid) used to make the MMAB enzyme. Researchers believe that nearly all of these mutations lead to the production of a nonfunctional version of the enzyme. As a result, AdoCbl cannot be made properly. A lack of AdoCbl impairs the function of methylmalonyl CoA mutase, which results in the incomplete breakdown of certain proteins and lipids. This defect allows toxic compounds to build up in the body's organs and tissues, causing the signs and symptoms of methylmalonic acidemia.

Chromosomal Location

Cytogenetic Location: 12q24.11, which is the long (q) arm of chromosome 12 at position 24.11

Molecular Location: base pairs 109,553,715 to 109,573,553 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- ATP:Cob(I)alamin Adenosyltransferase
- ATR
- cblB
- CFAP23
- cob(I)alamin adenosyltransferase
- methylmalonic aciduria (cobalamin deficiency) type B
- MMAB_HUMAN

Additional Information & Resources

Educational Resources

- Basic Neurochemistry (sixth edition, 1999): Methylmalonic aciduria may be secondary to defects of cobalamin metabolism https://www.ncbi.nlm.nih.gov/books/NBK27933/#A3118

GeneReviews

- Isolated Methylmalonic Acidemia https://www.ncbi.nlm.nih.gov/books/NBK1231

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MMAB%5BTIAB%5D%29+OR+%28cblB%5BTIAB%5D%29+AND+%28methylmalonic%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- MMAB GENE
  http://omim.org/entry/607568

Research Resources

- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MMAB%5Bgene%5D
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=19331
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/326625
- UniProt
  http://www.uniprot.org/uniprot/Q96EY8

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